result in loss of user privileges and other penalties.

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FILE 'HOME' ENTERED AT 13:06:12 ON 09 MAR 2005

=> file .biotech
COST IN U.S. DOLLARS

	SINCE FILE ENTRY	TOTAL SESSION
FULL ESTIMATED COST	0.21	0.21

FILE 'MEDLINE' ENTERED AT 13:06:34 ON 09 MAR 2005

FILE 'BIOSIS' ENTERED AT 13:06:34 ON 09 MAR 2005
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FILE 'BIOTECHDS' ENTERED AT 13:06:34 ON 09 MAR 2005
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FILE 'CAPLUS' ENTERED AT 13:06:34 ON 09 MAR 2005
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=> s ((individual? or personal?) medicine) (l) (genom? or gene or nucleic or DNA)
MISSING OPERATOR ERSONAL?) MEDICINE
The search profile that was entered contains terms or
nested terms that are not separated by a logical operator.

=> s ((individual? or personal?) (2a) medicine) (l) (genom? or gene or nucleic or DNA)
L1 425 ((INDIVIDUAL? OR PERSONAL?) (2A) MEDICINE) (L) (GENOM? OR GENE OR NUCLEIC OR DNA)

=> s sequenc? (l) l1
L2 116 SEQUENC? (L) L1

=> s health care
L3 592774 HEALTH CARE

=> s l2 and l3
L4 11 L2 AND L3

=> dup rem l4
PROCESSING COMPLETED FOR L4
L5 7 DUP REM L4 (4 DUPLICATES REMOVED)

=> d ibib abs l5 1-7

L5 ANSWER 1 OF 7 MEDLINE on STN DUPLICATE 1
ACCESSION NUMBER: 2005089387 IN-PROCESS
DOCUMENT NUMBER: PubMed ID: 15719589
TITLE: Moving toward whole-genome analysis: a technology perspective.
AUTHOR: Kreiner Thane; Buck Katie Tillman
CORPORATE SOURCE: Affymetrix, Inc., Santa Clara, CA 95051, USA.. thane_kreiner@affymetrix.com
SOURCE: American journal of health-system pharmacy : AJHP : official journal of the American Society of Health-System Pharmacists, (2005 Feb 1) 62 (3) 296-305. Journal code: 9503023. ISSN: 1079-2082.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: NONMEDLINE; IN-PROCESS; NONINDEXED; Health; Priority Journals

ENTRY DATE: Entered STN: 20050222
Last Updated on STN: 20050222

AB PURPOSE: New, highly efficient technologies used in **genomic** analysis are described, and their implications for **health care** are discussed. SUMMARY: The availability of the human **genome sequence**, in confluence with the ability to affordably package it for analysis, is opening new frontiers in biomedical research. On the horizon, **personalized medicine** --driven by molecular characterization of disease, genetic analysis of the patient, and information technologies designed to enable **health care** professionals to leverage these tools--promises to fundamentally transform **health care**. New genetics technologies, such as high-density microarrays, will fuel this research by providing researchers with the ability to comprehensively access the human **genome** in all its complexity. Some of the most promising areas for application of genetic information are those where society's current needs are greatest: complex, common disorders, such as cancer and cardiovascular disease; drug interactions; inherited genetic disorders that afflict children; and late-onset conditions for which no cure currently exists. The barriers to using genetic information widely in **health care** are in many cases not technological or economic, but social and political. CONCLUSION: New technology enables efficient, large-scale analysis of the whole **genome**, genetic variations, and **gene** expression. **Genomic** analysis has profound clinical, economic, and social implications for **health care**.

L5 ANSWER 2 OF 7 CAPLUS COPYRIGHT 2005 ACS on STN
ACCESSION NUMBER: 2004:582945 CAPLUS
DOCUMENT NUMBER: 141:224792
TITLE: Genomic Messaging System and DNA Mark-Up Language for Information-Based Personalized Medicine with Clinical and Proteome Research Applications
AUTHOR(S): Robson, Barry; Mushlin, Richard
CORPORATE SOURCE: T.J. Watson Research Lab., IBM Research, Yorktown Heights, NY, 10598, USA
SOURCE: Journal of Proteome Research (2004), 3(5), 930-948
CODEN: JPROBS; ISSN: 1535-3893
PUBLISHER: American Chemical Society
DOCUMENT TYPE: Journal
LANGUAGE: English

AB The convergence of clin. medicine and the Life Sciences, commencing with opportunities in clin. trials and clin. linked medical research, presents many novel challenges. The Genomic Messaging System (GMS) described here was originally developed as a tool for assembling clin. genomic records of individual and collective patients, and was then generalized to become a flexible work-flow component that will link clin. records to a variety of computational biol. research tools, for research and ultimately for a more personalized, focused, and preventative **health-care** system. Prominent among the applications linked are protein science applications, including the rapid automated modeling of patient proteins with their individual structural polymorphisms. In an initial study, GMS formed the basis of a fully automated system for modeling patient proteins with structural polymorphisms as a basis for drug selection and ultimately design on an individual patient basis.

REFERENCE COUNT: 17 THERE ARE 17 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L5 ANSWER 3 OF 7 CAPLUS COPYRIGHT 2005 ACS on STN DUPLICATE 2
ACCESSION NUMBER: 2005:11255 CAPLUS
TITLE: Applying pharmacogenomics in drug development: Call for collaborative efforts
AUTHOR(S): Gurwitz, David
CORPORATE SOURCE: Department of Human Genetics and Molecular Medicine, Sackler Faculty of Medicine, Tel-Aviv University, Tel-Aviv, Israel
SOURCE: Drug Development Research (2004), 62(2), 71-75
CODEN: DDREDK; ISSN: 0272-4391

PUBLISHER: Wiley-Liss, Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
AB **Personalized medicine** remains the long-awaited next revolution in medicine. So far, progress towards this change has been much slower than hoped, given that our entire **DNA sequence** has been publicly available since Apr. 2001. Three years down the road, it has become clear that the expectations for fast progress in medicine were excessive, and that our **genome** is by far more complex than originally perceived. Moreover, it seems that both the medical profession and **health care** systems, as well as pharmaceutical companies, are too conservative for modifying diagnostic and treatment protocols following the gain of new pharmacogenomics knowledge that has the potential to drastically reduce the incidence rates of adverse drug reactions. The next few years may well be a crucial turning point for the use of pharmacogenomics data in drug development. The transformation will hopefully begin with the availability of FDA approved rapid and reliable diagnostic screening tools for CYP450 alleles, along with new FDA guidelines favoring the approval process for drug applications supported by valuable **genomic** information related to toxic reactions. The current theme issue, a focused snapshot for mid-2004, highlights some of the vital topics in clin. genetics and informatics that together will hopefully form a platform for future joint efforts to identify the most valuable genotype/drug response phenotype correlations. Dialogue and collaboration between regulatory agencies and the pharmaceutical industry, as well as within the pharmaceutical sector, will be indispensable for advancing beyond this turning point towards genuine **personalized medicine**.
REFERENCE COUNT: 25 THERE ARE 25 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L5 ANSWER 4 OF 7 MEDLINE on STN DUPLICATE 3
ACCESSION NUMBER: 2003591245 MEDLINE
DOCUMENT NUMBER: PubMed ID: 14672519
TITLE: Race, distributive justice and the promise of pharmacogenomics: ethical considerations.
AUTHOR: Lee Sandra Soo-Jin
CORPORATE SOURCE: Center for Biomedical Ethics, Stanford University Medical School and the Department of Cultural and Social Anthropology, Stanford University, Stanford, California 94304-1703, USA.. sandra.lee@stanford.edu
CONTRACT NUMBER: K01 HL72465 (NHLBI)
SOURCE: American journal of pharmacogenomics : genomics-related research in drug development and clinical practice, (2003) 3 (6) 385-92. Ref: 38
Journal code: 100967746. ISSN: 1175-2203.
PUB. COUNTRY: New Zealand
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
(REVIEW, TUTORIAL)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200402
ENTRY DATE: Entered STN: 20031216
Last Updated on STN: 20040302
Entered Medline: 20040227
AB Pharmacogenomics has emerged in the popular press as a key vehicle ushering in a new era of **personalized medicine**. Often described in utopian terms, **gene-sequencing** technology is predicted to result in the creation of a new line of therapeutics tailored to individual genetic signatures. In the absence of cost-effective, ubiquitous **genome** scanning tests, it may be more accurate to describe the next wave of **genomic** medicine as population-based rather than one focused on individual differences. Although the completion of the Human **Genome** Project seemed to confirm the fallacy of a genetic basis of 'race', the use of race in understanding human genetic variation has become a central focal point in the development of tools in **genomic** research in medicine.

Despite the often repeated statement that humans share 99.9% of their genetic makeup, the growing number of privately and publicly funded cell repositories collecting **DNA** samples from racially identified populations reflects the increasing salience of the relationship between race and genes.Research on the ethical implications of identifying race in pharmacogenomics research has thus far, been fairly limited. As the field surges ahead, it is critical to examine the use of race in pharmacogenomics research and its attendant benefits and potential harm to individuals and groups.

L5 ANSWER 5 OF 7 EMBASE COPYRIGHT 2005 ELSEVIER INC. ALL RIGHTS RESERVED. on STN
ACCESSION NUMBER: 2003345723 EMBASE
TITLE: The human genome and the future of medicine.
AUTHOR: Mattick J.S.
CORPORATE SOURCE: Prof. Dr. J.S. Mattick, Institute for Molecular Bioscience, University of Queensland, St. Lucia, QLD 4072, Australia. j.mattick@imb.uq.edu.au
SOURCE: Medical Journal of Australia, (18 Aug 2003) 179/4 (212-216).
Refs: 28
ISSN: 0025-729X CODEN: MJAUAJ
COUNTRY: Australia
DOCUMENT TYPE: Journal; General Review
FILE SEGMENT: 022 Human Genetics
029 Clinical Biochemistry
005 General Pathology and Pathological Anatomy
036 Health Policy, Economics and Management
037 Drug Literature Index
004 Microbiology
030 Pharmacology
038 Adverse Reactions Titles
017 Public Health, Social Medicine and Epidemiology
LANGUAGE: English
SUMMARY LANGUAGE: English
AB The draft human **genome sequence** (about 3 billion base pairs) was completed in 2001. Humans have fewer protein-coding genes than expected, and most of these are highly conserved among animals. Humans and other complex organisms produce massive amounts of non-coding RNAs, which may form another level of genetic output that controls differentiation and development. Aside from classical monogenic diseases and other differences caused by mutations and polymorphisms in protein-coding genes, much of the variation between individuals, including that which may affect our predispositions to common diseases, is probably due to differences in the non-coding regions of the **genome** (ie, the control architecture of the system). Within 10 years we can expect to see: • increased penetration of DMA diagnostic tests to assess risk of disease, to diagnose pathogens, to determine the best treatment regimens, and for individual identification; • a range of new Pharmaceuticals as well as new **gene** and cell therapies to repair damage, to optimise health and to minimise future disease risk; and • **medicine** become increasingly **personalised**, with the knowledge of individual genetic make-up and lifestyle influences.

L5 ANSWER 6 OF 7 CAPLUS COPYRIGHT 2005 ACS on STN DUPLICATE 4
ACCESSION NUMBER: 2002:386123 CAPLUS
DOCUMENT NUMBER: 137:319793
TITLE: Pharmacogenetic applications of the postgenomic era
AUTHOR(S): Sengupta, L. K.; Sengupta, Susmita; Sarkar, Munna
CORPORATE SOURCE: Department of Genetics, Barkatullah University, Bhopal, 462 026, India
SOURCE: Current Pharmaceutical Biotechnology (2002), 3(2), 141-150
CODEN: CPBUBP; ISSN: 1389-2010
PUBLISHER: Bentham Science Publishers Ltd.
DOCUMENT TYPE: Journal; General Review
LANGUAGE: English
AB A review. The human **genome sequence** has provided a

view of the internal genetic scaffold around which human life is molded. We have inherited this heritage from our ancestors and through it we are connected to all life on earth. The **sequencing** of the human **genome**, among other things, has led to newer areas of **health care** and medicine. The human population is heterogeneous and consists of populations of immense ethnic diversity. There are considerable allelic differences among human populations as well as individuals within each ethnic group as a result of the mol. heterogeneity of the **genome**. This, in turn, is responsible for differential allelic expression of genes, endowing them with polymorphic characters. The mol. diversity within genes is responsible, among other things, for disease resistance or susceptibility or drug response. This review discusses nuances of the genetic repertoire and correlates these with identification of disease **gene**, genes that have been or can be used as drug targets, and candidate genes for drug development, as well as recent trends in drug discovery. As regular clin. trials of drugs do not take into account ethnic variations, there are sometimes differential responses with respect to the efficacy of and/or adverse reactions to a drug. Therefore the diverse ethnic populations of the world pose a challenge to the pharmaceutical industry. The concept of **personal medicine** seems to be the answer to this problem. However, this is a Herculean task, requiring immense innovation in technol.; it would be time consuming and is not a financially viable proposition at this time. An alternate approach would be to divide populations into genetic cohorts and design drugs according to their genetic profile and haplotype. In addition, ethical and legal considerations must also be taken into consideration.

REFERENCE COUNT: 65 THERE ARE 65 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L5 ANSWER 7 OF 7 EMBASE
on STN

ACCESSION NUMBER: 2001439026 EMBASE

TITLE: Fruits of human genome project and private venture, and their impact on life science.

AUTHOR: Ikekawa A.; Ikekawa S.

CORPORATE SOURCE: A. Ikekawa, 1-13-2, Mihamaku, Makuhari-Nishi, Chiba 261-0026, Japan

SOURCE: Yakugaku Zasshi, (2001) 121/12 (845-873).
Refs: 154
ISSN: 0031-6903 CODEN: YKKZAJ

COUNTRY: Japan

DOCUMENT TYPE: Journal; General Review

FILE SEGMENT: 017 Public Health, Social Medicine and Epidemiology
022 Human Genetics
039 Pharmacy

LANGUAGE: English

SUMMARY LANGUAGE: English; Japanese

AB A small knowledge base was created by organizing the Human **Genome** Project (HGP) and its related issues in "Science" magazines between 1996 and 2000. This base revealed the stunning achievement of HGP and a private venture and its impact on today's biology and life science. In the mid-1990, they encouraged the developement of advanced high throughput automated **DNA sequencers** and the technologies that can analyse all genes at once in a systematic fashion. Using these technologies, they completed the **genome sequence** of human and various other organisms. These fruits opened the door to comparative **genomics**, functional **genomics**, the interdisprinary field between computer and biology, and proteomics. They have caused a shift in biological investigation from studying single genes or proteins to studying all genes or proteins at once, and causing revolutional changes in traditional biology, drug discovery and therapy. They have expanded the range of potential drug targets and have facilitated a shift in drug discovery programs toward rational target-based strategies. They have spawned pharmacogenomics that could give rise to a new generation of highly effective drugs that treat causes, not just symptoms. They should also cause a migration from the traditional medications that are safe and effective for every members of the

population to **personalised medicine** and
personalised therapy.

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=> FIL STNGUIDE
COST IN U.S. DOLLARS                                SINCE FILE      TOTAL
                                                       ENTRY    SESSION
FULL ESTIMATED COST                                    40.11      40.32

DISCOUNT AMOUNTS (FOR QUALIFYING ACCOUNTS)          SINCE FILE      TOTAL
                                                       ENTRY    SESSION
CA SUBSCRIBER PRICE                                    -2.19      -2.19

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FILE CONTAINS CURRENT INFORMATION.
LAST RELOADED: Mar  4, 2005 (20050304/UP).

=> file .biotech
COST IN U.S. DOLLARS                                SINCE FILE      TOTAL
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FULL ESTIMATED COST                                     0.42      40.74

DISCOUNT AMOUNTS (FOR QUALIFYING ACCOUNTS)          SINCE FILE      TOTAL
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CA SUBSCRIBER PRICE                                     0.00      -2.19

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     FILE 'MEDLINE, BIOSIS, BIOTECHDS, CAPLUS, EMBASE' ENTERED AT 13:06:34 ON
     09 MAR 2005
L1          425 S ((INDIVIDUAL? OR PERSONAL?) (2A) MEDICINE) (L) (GENOM? OR GEN
L2          116 S SEQUENC? (L) L1
L3       592774 S HEALTH CARE
L4           11 S L2 AND L3
L5            7 DUP REM L4 (4 DUPLICATES REMOVED)

     FILE 'STNGUIDE' ENTERED AT 13:12:46 ON 09 MAR 2005

     FILE 'MEDLINE, BIOSIS, BIOTECHDS, CAPLUS, EMBASE' ENTERED AT 13:17:10 ON
     09 MAR 2005

=> s health information
L6         7344 HEALTH INFORMATION

=> s l6 and l2
L7            0 L6 AND L2
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=> s l2 and l3
L8          11 L2 AND L3

=> s health care information
L9         937 HEALTH CARE INFORMATION

=> s l9 and l2
L10          0 L9 AND L2

=> s search? and archiv?
L11       1078 SEARCH? AND ARCHIV?

=> s (search? or  archiv?) (l) l2
L12          4 (SEARCH? OR ARCHIV?) (L) L2

=> d ibib l12 1-4

L12  ANSWER 1 OF 4     MEDLINE on STN
ACCESSION NUMBER:   2003465618     MEDLINE
DOCUMENT NUMBER:    PubMed ID: 14527308
TITLE:              Nonsyndromic seizure disorders: epilepsy and the use of the
                    internet to advance research.
AUTHOR:             Leppert Mark F; Singh Nanda A
CORPORATE SOURCE:   Department of Human Genetics, University of Utah, Salt Lake
                    City, Utah 84112-5330, USA.. mleppert@genetics.utah.edu
SOURCE:             Annual review of genomics and human genetics, (2003) 4
                    437-57.  Ref: 59
                    Journal code: 100911346. ISSN: 1527-8204.
PUB. COUNTRY:       United States
DOCUMENT TYPE:      Journal; Article; (JOURNAL ARTICLE)
                    General Review; (REVIEW)
                    (REVIEW, TUTORIAL)
LANGUAGE:           English
FILE SEGMENT:       Priority Journals
ENTRY MONTH:        200312
ENTRY DATE:         Entered STN: 20031008
                    Last Updated on STN: 20031218
                    Entered Medline: 20031203

L12  ANSWER 2 OF 4     MEDLINE on STN
ACCESSION NUMBER:   2002105502     MEDLINE
DOCUMENT NUMBER:    PubMed ID: 11837493
TITLE:              The human genome project: implications for the
                    endocrinologist.
AUTHOR:             Francke U
CORPORATE SOURCE:   Department of Genetics, Beckman Center for Molecular and
                    Genetic Medicine, Stanford University School of Medicine,
                    CA 94305-5323, USA.. francke@cmgm.stanford.edu
SOURCE:             Journal of pediatric endocrinology & metabolism : JPEM,
                    (2001) 14 Suppl 6 1395-408.  Ref: 30
                    Journal code: 9508900. ISSN: 0334-018X.
PUB. COUNTRY:       England: United Kingdom
DOCUMENT TYPE:      Journal; Article; (JOURNAL ARTICLE)
                    General Review; (REVIEW)
                    (REVIEW, TUTORIAL)
LANGUAGE:           English
FILE SEGMENT:       Priority Journals
ENTRY MONTH:        200207
ENTRY DATE:         Entered STN: 20020212
                    Last Updated on STN: 20020720
                    Entered Medline: 20020719

L12  ANSWER 3 OF 4  CAPLUS  COPYRIGHT 2005 ACS on STN
ACCESSION NUMBER:       2003:911580  CAPLUS
DOCUMENT NUMBER:        140:88182
TITLE:                  Nonsyndromic seizure disorders: Epilepsy and the use
                        of the internet to advance research
```

AUTHOR(S): Leppert, Mark F.; Singh, Nanda A.
CORPORATE SOURCE: Department of Human Genetics, University of Utah, Salt Lake City, UT, 84112-5330, USA
SOURCE: Annual Review of Genomics and Human Genetics (2003), 4, 437-457, 2 plates
CODEN: ARGHC4; ISSN: 1527-8204
PUBLISHER: Annual Reviews Inc.
DOCUMENT TYPE: Journal; General Review
LANGUAGE: English
REFERENCE COUNT: 59 THERE ARE 59 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L12 ANSWER 4 OF 4 EMBASE COPYRIGHT 2005 ELSEVIER INC. ALL RIGHTS RESERVED.
on STN
ACCESSION NUMBER: 2003461588 EMBASE
TITLE: Nonsyndromic Seizure Disorders: Epilepsy and the Use of the Internet to Advance Research.
AUTHOR: Leppert M.F.; Singh N.A.
CORPORATE SOURCE: M.F. Leppert, Department of Human Genetics, University of Utah, Salt Lake City, UT 84112-5330, United States. mleppert@genetics.utah.edu
SOURCE: Annual Review of Genomics and Human Genetics, (2003) 4/- (437-457).
Refs: 59
ISSN: 1527-8204 CODEN: ARGHC4
COUNTRY: United States
DOCUMENT TYPE: Journal; General Review
FILE SEGMENT: 008 Neurology and Neurosurgery
022 Human Genetics
050 Epilepsy
LANGUAGE: English
SUMMARY LANGUAGE: English

=> FIL STNGUIDE

COST IN U.S. DOLLARS	SINCE FILE ENTRY	TOTAL SESSION
FULL ESTIMATED COST	30.34	71.08

DISCOUNT AMOUNTS (FOR QUALIFYING ACCOUNTS)	SINCE FILE ENTRY	TOTAL SESSION
CA SUBSCRIBER PRICE	0.00	-2.19

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FILE CONTAINS CURRENT INFORMATION.
LAST RELOADED: Mar 4, 2005 (20050304/UP).

=> d his

(FILE 'HOME' ENTERED AT 13:06:12 ON 09 MAR 2005)

FILE 'MEDLINE, BIOSIS, BIOTECHDS, CAPLUS, EMBASE' ENTERED AT 13:06:34 ON 09 MAR 2005
L1 425 S ((INDIVIDUAL? OR PERSONAL?) (2A) MEDICINE) (L) (GENOM? OR GEN
L2 116 S SEQUENC? (L) L1
L3 592774 S HEALTH CARE
L4 11 S L2 AND L3
L5 7 DUP REM L4 (4 DUPLICATES REMOVED)

FILE 'STNGUIDE' ENTERED AT 13:12:46 ON 09 MAR 2005

FILE 'MEDLINE, BIOSIS, BIOTECHDS, CAPLUS, EMBASE' ENTERED AT 13:17:10 ON 09 MAR 2005
L6 7344 S HEALTH INFORMATION

```
L7            0 S L6 AND L2
L8           11 S L2 AND L3
L9          937 S HEALTH CARE INFORMATION
L10           0 S L9 AND L2
L11        1078 S SEARCH? AND ARCHIV?
L12           4 S (SEARCH? OR  ARCHIV?) (L) L2

     FILE 'STNGUIDE' ENTERED AT 13:22:19 ON 09 MAR 2005

=> dup rem l2
COST IN U.S. DOLLARS                                SINCE FILE      TOTAL
                                                      ENTRY    SESSION
FULL ESTIMATED COST                                    0.78      71.86

DISCOUNT AMOUNTS (FOR QUALIFYING ACCOUNTS)          SINCE FILE      TOTAL
                                                      ENTRY    SESSION
CA SUBSCRIBER PRICE                                    0.00      -2.19

FILE 'MEDLINE' ENTERED AT 13:30:12 ON 09 MAR 2005

FILE 'BIOSIS' ENTERED AT 13:30:12 ON 09 MAR 2005
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PROCESSING COMPLETED FOR L2
L13          54 DUP REM L2 (62 DUPLICATES REMOVED)

=> s l13 not py>2001
L14          13 L13 NOT PY>2001

=> d ibib abs l14 1-13

L14  ANSWER 1 OF 13     MEDLINE on STN
ACCESSION NUMBER:   2002105502     MEDLINE
DOCUMENT NUMBER:    PubMed ID: 11837493
TITLE:              The human genome project: implications for the
                    endocrinologist.
AUTHOR:             Francke U
CORPORATE SOURCE:   Department of Genetics, Beckman Center for Molecular and
                    Genetic Medicine, Stanford University School of Medicine,
                    CA 94305-5323, USA.. francke@cmgm.stanford.edu
SOURCE:             Journal of pediatric endocrinology & metabolism : JPEM,
                    (2001) 14 Suppl 6 1395-408.  Ref: 30
                    Journal code: 9508900. ISSN: 0334-018X.
PUB. COUNTRY:       England: United Kingdom
DOCUMENT TYPE:      Journal; Article; (JOURNAL ARTICLE)
                    General Review; (REVIEW)
                    (REVIEW, TUTORIAL)
LANGUAGE:           English
FILE SEGMENT:       Priority Journals
ENTRY MONTH:        200207
ENTRY DATE:         Entered STN: 20020212
                    Last Updated on STN: 20020720
                    Entered Medline: 20020719
AB   The sequencing of the human genome is a major
     achievement of our time.  This article reviews the process and current
     status of the working draft sequence, ways to predict genes and
     assign function, and conclusions for human biology.  Gene
     density is uneven and related to chromosome banding patterns, and the
```

estimate of approximately 30,000 genes is lower than expected. Genetic maps for men and women differ from each other and from the physical map. Single nucleotide polymorphisms occur at an average spacing of 1 kb. Human populations are 99.99% identical, and most **sequences** are shared between people from different continents. To illustrate the tools for accessing the human **genome sequence**, searches were performed for genes encoding three categories of growth-related proteins, insulin-like growth factor-I (IGF-I) receptor, IGF-binding proteins and growth hormone receptor. The results revealed novel details about their **genomic** organization and new predicted transcripts. Impacts on medicine are promised in the fields of diagnostics (development of new tests), therapeutics (identification of new potential drug targets) and pharmacogenomics (streamlining of drug discovery and **personalized medicine**). Associated ethical, legal and social implications and controversies include genetic determinism, informed consent, privacy and confidentiality, ownership of genetic information in the biotechnology marketplace, and access to genetic healthcare.

L14 ANSWER 2 OF 13 MEDLINE on STN
ACCESSION NUMBER: 2002039820 MEDLINE
DOCUMENT NUMBER: PubMed ID: 11766401
TITLE: Fruits of human genome project and private venture, and their impact on life science.
AUTHOR: Ikekawa A; Ikekawa S
SOURCE: Yakugaku zasshi. Journal of the Pharmaceutical Society of Japan, (2001 Dec) 121 (12) 845-73. Ref: 131
Journal code: 0413613. ISSN: 0031-6903.
PUB. COUNTRY: Japan
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
(REVIEW, TUTORIAL)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200201
ENTRY DATE: Entered STN: 20020124
Last Updated on STN: 20020125
Entered Medline: 20020115

AB A small knowledge base was created by organizing the Human **Genome** Project (HGP) and its related issues in "Science" magazines between 1996 and 2000. This base revealed the stunning achievement of HGP and a private venture and its impact on today's biology and life science. In the mid-1990, they encouraged the development of advanced high throughput automated **DNA sequencers** and the technologies that can analyse all genes at once in a systematic fashion. Using these technologies, they completed the **genome sequence** of human and various other organisms. These fruits opened the door to comparative **genomics**, functional **genomics**, the interdisprinary field between computer and biology, and proteomics. They have caused a shift in biological investigation from studying single genes or proteins to studying all genes or proteins at once, and causing revolutional changes in traditional biology, drug discovery and therapy. They have expanded the range of potential drug targets and have facilitated a shift in drug discovery programs toward rational target-based strategies. They have spawned pharmacogenomics that could give rise to a new generation of highly effective drugs that treat causes, not just symptoms. They should also cause a migration from the traditional medications that are safe and effective for every members of the population to **personalized medicine** and **personalized** therapy.

L14 ANSWER 3 OF 13 MEDLINE on STN
ACCESSION NUMBER: 2002012668 MEDLINE
DOCUMENT NUMBER: PubMed ID: 11474566
TITLE: [New progress and new tools for the study of molecular genetics in dyslipoproteinemia].
Nouveaux progres et nouveaux outils d'etude de la genetique moleculaire des dyslipoproteinemies.
AUTHOR: Benlian P

CORPORATE SOURCE: Praticien Hospitalier, Laboratoire de Biochimie et de Biologie Moleculaire, Hopital Saint Antoine, 184 rue du faubourg Saint Antoine-75012 Paris.
SOURCE: Bulletin de l'Academie nationale de medecine, (2001) 185 (1) 21-31; discussion 32-3.
Journal code: 7503383. ISSN: 0001-4079.
PUB. COUNTRY: France
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: French
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200112
ENTRY DATE: Entered STN: 20020121
Last Updated on STN: 20020121
Entered Medline: 20011205

AB More than three centuries after Mendel, at the era of electronic and computed information taking over the principle of information transmitted in discrete "packets" on the "internet", the **sequence** of the human **genome** is about to be completely released on public databases accessible on that very same internet. The **gene**, classically a virtual object, has become after several decades of intensive progress in cellular and molecular biology, a real object commonly manipulated and analyzed. More than fifty genes have been identified in the regulation of lipoprotein metabolism, giving rise to novel molecular pathophysiological bases for dyslipoproteinemia and beyond to other disorders related with lipid homeostasis. Dyslipoproteinemia, or disorders of lipoprotein metabolism commonly considered as lifestyle and age-related diseases, have now a molecular basis. Novel clinical entities no longer defined as "essential", but as molecular-based are progressively individualized. Novel tools for the diagnosis, prognosis or treatment have already modified the way these silent and frequent diseases are managed in clinical practice. In that respect, dyslipoproteinemia are among pioneer diseases in the medicine of the new millennium, which progressively evolves from a fact-based **medicine** to the **individualized** prevention of morbid events.

L14 ANSWER 4 OF 13 MEDLINE on STN
ACCESSION NUMBER: 2001322862 MEDLINE
DOCUMENT NUMBER: PubMed ID: 11307306
TITLE: The present status and future prospect of the molecular diagnostic tests.
AUTHOR: Miyachi H
CORPORATE SOURCE: Department of Laboratory Medicine, Tokai University School of Medicine, Isehara 259-1193.
SOURCE: Rinsho byori. Japanese journal of clinical pathology, (2001 Feb) 49 (2) 139-49. Ref: 49
Journal code: 2984781R. ISSN: 0047-1860.
PUB. COUNTRY: Japan
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
LANGUAGE: Japanese
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200106
ENTRY DATE: Entered STN: 20010611
Last Updated on STN: 20010611
Entered Medline: 20010607

AB Assays for **DNA** or RNA **sequences** to diagnose infectious, neoplastic and genetic diseases have been widely used through recent progress in the molecular biology and biotechnology, and are now essential in care of patients under the advanced medicine through earlier and more accurate diagnosis. Automated systems have been developed for amplification and detection of **nucleic** acid **sequence** for infectious agents, using various **nucleic** acid amplification technology such as PCR. A fully automated PCR system and automated extraction of specific **sequence** for infectious agents such as hepatitis C virus RNA has been developed. These automated systems have provided improvement of not only assay efficiency but also quality control of the tests and have contributed to the standardization of them. Importance of development of systems for quality assessment and laboratory

accreditation has been emphasized, particularly in those that still have been performed with manual methods. Based on the information on the **genome sequence** as the outcome of the human **genome** project, functions of genes and proteins have been studied by post-**genomics** such as expression profiling using **DNA** microarray, proteomics, single nucleotide polymorphisms analysis, coupled with bioinformatics. Along with advances in pharmacogenomics, these studies have raised the prospect of the development of tests for **individualized medicine** based on genetic information such as those predicting individual susceptibility to diseases for prevention and responsiveness to drugs for choice of treatment. For practice of such medicine, each genetic information and tests for it must be carefully evaluated and determined whether it is appropriate for cost-effective medicine through contributions to efficient process of decision-makings on patient care for prevention or avoidance of diseases and thus to cost savings.

L14 ANSWER 5 OF 13 MEDLINE on STN
ACCESSION NUMBER: 2001228253 MEDLINE
DOCUMENT NUMBER: PubMed ID: 11291222
TITLE: A revolution in genetics: changing medicine, changing lives.
AUTHOR: Bottles K
CORPORATE SOURCE: Genomics Collaborative, Inc., Cambridge, Massachusetts, USA.. kbottles@genecoop.com
SOURCE: Physician executive, (2001 Mar-Apr) 27 (2) 58-63. Journal code: 8610398. ISSN: 0898-2759.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Health
ENTRY MONTH: 200104
ENTRY DATE: Entered STN: 20010502
Last Updated on STN: 20010502
Entered Medline: 20010426

AB The **sequencing** of the human **genome** is only the tip of the iceberg. It is the beginning of a revolution that many predict will transform medicine. How will genetics research affect physicians and patients and the practice of medicine? When investigators identify the function and association of human genes with common chronic diseases, diagnosis, treatment, and classification of human diseases will be changed forever. Genetic susceptibility testing allows patients to know their predisposition to disease long before symptoms appear. Physicians can intervene with customized advice so that the patient can prevent, modify, or avoid the predisposed condition by better understanding both his or her genetic and environmental risk for disease. The promise of a genetic approach to drug therapy involves moving from one size fits all to **personalized medicine** tailored to the individual patient. Physicians will become mentors and counselors, advising patients on the best treatment path given their unique genetic predisposition--even in this sophisticated, high tech field, the physician-patient relationship is likely to improve, highlighted by individualized therapies and personal attention.

L14 ANSWER 6 OF 13 MEDLINE on STN
ACCESSION NUMBER: 2001164423 MEDLINE
DOCUMENT NUMBER: PubMed ID: 11265659
TITLE: [From genomics to therapeutics]. De la genomique a la therapeutique.
AUTHOR: Kahn A
CORPORATE SOURCE: Institut Cochin de Genetique Moleculaire, Unite 129 de l'Institut National de la Sante et de la Recherche Medicale, CHU Cochin Port-Royal, 24, rue du Faubourg Saint Jacques-75014 Paris.
SOURCE: Bulletin de l'Academie nationale de medecine, (2000) 184 (7) 1463-75; discussion 1475-6. Ref: 36 Journal code: 7503383. ISSN: 0001-4079.
PUB. COUNTRY: France

DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
(REVIEW, TUTORIAL)
LANGUAGE: French
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200106
ENTRY DATE: Entered STN: 20010611
Last Updated on STN: 20010611
Entered Medline: 20010607

AB The **sequencing** of the human **genome** will be achieved in the first years of the next century. This program is often presented as constituting a huge hope for medicine. It is sometimes expected that a disease-free world is a realistic prospect for tomorrow. Otherwise, the future of therapeutics is viewed as characterized by a **personalized medicine** in which each person will be preventively or curatively treated in function of its genetic make-up, assuring a maximal efficacy and the absence of toxicity. In fact, we have to be cautious, even if, indeed, progress are expected in chemotherapy and biological therapies. Expected difficulties will arise from the nature of the main diseases persisting in the developed countries, and from the economical situation in the developing ones. Fortunately for the modern doctors, their grand-grand children will still have the possibility to become themselves doctors: patients requiring treatments will still be there!

L14 ANSWER 7 OF 13 MEDLINE on STN
ACCESSION NUMBER: 2000297250 MEDLINE
DOCUMENT NUMBER: PubMed ID: 10838758
TITLE: [The method of DNA isolation can affect the rate of preferential amplification of alleles by the polymerase chain reaction].
Metoda izolace DNA muze ovlivnit stupen preferencni amplifikace alel polymerazovou retezovou reakci.
AUTHOR: Korabecna M
CORPORATE SOURCE: Biologicky ustav LF UK, Plzen.
SOURCE: Soudni lekarstvi / casopis Sekce soudniho lekarstvi Cs. lekarske spolecnosti J. Ev. Purkyne, (2000 Jan) 45 (1) 2-5.
Journal code: 9601665. ISSN: 0371-1854.
PUB. COUNTRY: Czech Republic
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: Czech
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200007
ENTRY DATE: Entered STN: 20000728
Last Updated on STN: 20000728
Entered Medline: 20000720

AB Today, polymerase chain reaction is a common part of approaches serving for identification of **individuals** in legal **medicine**. This method is easily practicable, however attention must be paid to the optimization of reaction conditions and to the interpretation of results. From the literature, such cases are known, in which during amplification of extremely small amount of **DNA** (e.g. from one cell) the polymerase chain reaction preferably amplifies only one of two in the template **DNA** present alleles. If the amplified fragments differ in length, the shorter one is amplified preferably, and it may be cause of false results. In the presented study, **DNA** from 23 stains of male blood on different fabrics was isolated by two different methods (by treatment with proteinase K and boiling and by treatment with Chelex 100). The obtained **DNA** samples were amplified using primers, they are complementary to the amelogenin **gene sequences**. The system is suitable for sex determination, because amplification of the X-chromosomal **sequence** provides a fragment in length of 632 bp, amplification of the Y-chromosomal one a fragment in length of 443 bp. The isolation method based on proteinase K led in 17.38% of samples to the very intensive preferential amplification of the longer allele, and therefore to a false result. The isolation method based on Chelex 100 provided in all cases correct results with clearly recognizable preferential amplification of the shorter allele. The reported results

accentuate the meaning of choice of the appropriate isolation method, the need of accurate PCR optimization, and the careful interpretations of its outputs.

L14 ANSWER 8 OF 13 MEDLINE on STN
ACCESSION NUMBER: 2000058170 MEDLINE
DOCUMENT NUMBER: PubMed ID: 10590665
TITLE: Expanding scope and impact of services provided by clinical laboratory practice through molecular diagnostics.
AUTHOR: Miyachi H
CORPORATE SOURCE: Department of Clinical Pathology, Tokai University School of Medicine, Isehara.
SOURCE: Rinsho byori. Japanese journal of clinical pathology, (1999 Oct) 47 (10) 919-25. Ref: 30
Journal code: 2984781R. ISSN: 0047-1860.
PUB. COUNTRY: Japan
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
(REVIEW, TUTORIAL)
LANGUAGE: Japanese
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200002
ENTRY DATE: Entered STN: 20000229
Last Updated on STN: 20000229
Entered Medline: 20000211
AB Recent progress in molecular biology and biotechnology has facilitated assays for **DNA** or RNA **sequences** to diagnose infectious, neoplastic and genetic diseases. Many of the assays have been used clinically, and are now an essential part of patient care under advanced medicine. A clinical laboratory needs to ensure services for clinical needs such as provision of tests with required turnaround time and high quality as well as consulting practice for individual requests. For quality assurance of assays, it is particularly important to monitor clinical validation of the results by correlating them with the patient's status to prove clinically relevant. Staff need to be trained to become familiar with both molecular pathogenesis and technology so that they can provide informative tests with high quality. Along with advances in pharmacogenomics, the findings of the human **genome** project have raised the prospect of developing tests for **individualized medicine** based on genetic information such as those predicting individual susceptibility to diseases to facilitate prevention and indicate responsiveness to drugs for choice of treatment. Molecular diagnostic tests will contribute to an efficient process of decision making on patient care and result in cost savings through earlier and more accurate diagnosis. The scope and impact of services provided by clinical laboratory practice through molecular diagnostics will continue to expand in its integration into clinical practice.

L14 ANSWER 9 OF 13 MEDLINE on STN
ACCESSION NUMBER: 89368787 MEDLINE
DOCUMENT NUMBER: PubMed ID: 2672298
TITLE: [DNA fingerprints and hypervariable regions: genetic marker with many application potentials in medicine and biology]. DNS-"fingerprints" und hypervariable Regionen: genetische Marker mit zahlreichen Anwendungsmoglichkeiten in Medizin und Biologie.
AUTHOR: Fey M F
CORPORATE SOURCE: Institut fur Medizinische Onkologie der Universitat, Inselspital Bern.
SOURCE: Schweizerische medizinische Wochenschrift, (1989 Jun 10) 119 (23) 815-25. Ref: 56
Journal code: 0404401. ISSN: 0036-7672.
PUB. COUNTRY: Switzerland
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
(REVIEW, TUTORIAL)
LANGUAGE: German
FILE SEGMENT: Priority Journals

ENTRY MONTH: 198909
ENTRY DATE: Entered STN: 19900309
Last Updated on STN: 20020125
Entered Medline: 19890929

AB **DNA** polymorphisms are based on variations in the nucleotide **sequences** of the **DNA** within a given population and are transmitted from parents to offspring by Mendelian inheritance. Most of these mutations are phenotypically silent. Two different types of **DNA** polymorphisms are restriction fragment length polymorphisms and highly variable regions (HVRs), the latter with many different alleles at a given locus. Molecular probes for HVRs (or **DNA** minisatellites) can detect a great number of cross-hybridising fragments dispersed throughout the **genome**. The polymorphic patterns of these fragments are completely individual-specific, hence termed **DNA** "fingerprints". **DNA** "fingerprinting" has been shown to be a powerful tool for establishing family relationships, for example in paternity disputes, and for the positive identification of **individuals** in forensic **medicine**. The technique may be used to document marrow engraftment in patients who have undergone allogeneic bone marrow transplantation. **DNA** "fingerprinting" is a new method of assessing clonality in human tumours by identifying clonal somatic mutations in the tumour **DNA**. Cloning of individual **DNA** "fingerprint" fragments yields locus-specific HVR probes which, due to their high rate of heterozygosity, are ideal for linkage analysis and prenatal diagnosis in single **gene** disorders. This is exemplified by adult polycystic kidney disease, which has been found by a 3'alpha-globin-HVR probe to be closely linked to the alpha-globin-**gene** cluster on chromosome 16p. Locus-specific HVR probes have been used for the molecular diagnosis of clonal chromosomal deletions or loss of heterozygosity at particular loci in a large variety of tumours. These findings are the basis for the identification of anti-oncogenes or putative tumour-suppressor genes in the human **genome**.

L14 ANSWER 10 OF 13 BIOSIS COPYRIGHT (c) 2005 The Thomson Corporation on STN
ACCESSION NUMBER: 2002:40086 BIOSIS
DOCUMENT NUMBER: PREV200200040086
TITLE: Colon cancer and nutrigenetics: modifier genes.
Original Title: Cancer du colon et nutrigenetique: roles des genes modificateurs.
AUTHOR(S): Junien, Claudine [Reprint author]
CORPORATE SOURCE: INSERM UR 383, Groupe Hospitalier Necker - Enfants Malades, 149, rue de Sevres, 75015, Paris, France
SOURCE: Annales de Medecine Interne, (Septembre, 2001) Vol. 152, No. 5, pp. 337-351. print.
CODEN: AMDIBO. ISSN: 0003-410X.
DOCUMENT TYPE: Article
LANGUAGE: French
ENTRY DATE: Entered STN: 2 Jan 2002
Last Updated on STN: 25 Feb 2002

AB About 5% of colon cancer cases correspond to classic hereditary monogenic mendelian transmission involving at least 8 major genes of predisposition to this tumor. Genes with more moderate effects, in association with other genes can contribute to the occurrence of sporadic polygenic forms. These genes confer susceptibility to environmental factors and can play the role of aggravating or protective modifier genes in the different hereditary forms. Foods can interact with these genes and modulate their expression. Moreover **sequence** variations (polymorphisms) in these genes may also be responsible for slower or more rapid metabolism of nutrients leading to toxic or carcinogenic compounds. If some foods, or "pharmafoods" can have beneficial effects in some individuals with a particular subtype of the disease, others can be inefficient or even detrimental in patients with the same disease but with a different genetic origin or if the genetic background is different. Moreover tumorigenic processes are diverse. Tumor progression depends on genetic and environmental factors different from tumor initiation and on the site of the tumor along the colon tract. Interactions with the gut flora, the lymphoid system and specific features of growth of the colon mucosa are

also important parameters. Today with a formidable genetic knowledge arising from the **genome** project, new epidemiological data integrating the genetic data for multiple markers and a better knowledge of the tumorigenic processes involved, a new discipline is emerging. "Nutrigenetics" which is the study of hereditary basis of individual variations in response to foods opens for the oncoming decade the era of a **personalized** predictive **medicine** based on a nutrition adapted to the genetic make up of each of us.

L14 ANSWER 11 OF 13 CAPLUS COPYRIGHT 2005 ACS on STN
ACCESSION NUMBER: 2001:794192 CAPLUS
DOCUMENT NUMBER: 136:396422
TITLE: Significance of human gene sequence determination: draft of the human genome
AUTHOR(S): Sugano-Mizushima, Junko; Sugano, Sumio
CORPORATE SOURCE: Institute of Medical Science, University of Medicine, Japan
SOURCE: Saishin Igaku (2001), 56(Sept., Zokango), 2042-2052
CODEN: SAIGAK; ISSN: 0370-8241
PUBLISHER: Saishin Igakusha
DOCUMENT TYPE: Journal; General Review
LANGUAGE: Japanese
AB A review. Tracks of the human **genome sequencing** projects were reviewed and the efficient use of human **genome** draft **sequences** in functional **genomics** was discussed. Ensemble database was described as a draft-**sequence** database. Some approaches in functional **genomics** such as the full-length cDNA **sequencing** projects, SNP anal. and **gene** expression profiling were discussed. Application of the **sequence** information for establishing **individual**-based **medicine** was also briefly discussed.

L14 ANSWER 12 OF 13 CAPLUS COPYRIGHT 2005 ACS on STN
ACCESSION NUMBER: 2000:478266 CAPLUS
DOCUMENT NUMBER: 134:203103
TITLE: The human genome business today
AUTHOR(S): Brown, Kathryn
CORPORATE SOURCE: Alexandria, VA, USA
SOURCE: Scientific American (2000), 283(1), 50-55
CODEN: SCAMAC; ISSN: 0036-8733
PUBLISHER: Scientific American, Inc.
DOCUMENT TYPE: Journal; General Review
LANGUAGE: English
AB A review and discussion with 4 refs. on the economic, medical, and social effects of completing the **sequence** of the human **genome** . The **genome sequencing** strategies of the Human **Genome** Project and Cerera **Genomics** are reviewed, and then potential applications of this knowledge are discussed. Pharmacogenetics raises the possibility of **personalized** **medicine** - the ability to identify which person could be helped with which drug. **Genome sequencing** of model organisms, such as the mouse and fruit fly offer other avenues to find new drugs. However, legal issues, such as when **sequences** should be patented, and social issues, such as the possibility of genetic discrimination after genetic testing, also play a role in the use of the human **genome**.

L14 ANSWER 13 OF 13 EMBASE COPYRIGHT 2005 ELSEVIER INC. ALL RIGHTS RESERVED. on STN
ACCESSION NUMBER: 2000263479 EMBASE
TITLE: Genetic polymorphism of RhD-negative associated haplotypes in the Chinese.
AUTHOR: Jun Cai Lan; Chen Q.; Da Lin Wu; Ding H.; Dao Be Pong; Zhao T.
CORPORATE SOURCE: T. Zhao, Molec. and Cell. Immunogenetics Sec., NIAID, National Institutes of Health, 9000 Rockville Pike, Bethesda, MD 20892, United States. tzhao@niaid.nih.gov
SOURCE: Journal of Human Genetics, (2000) 45/4 (224-227).

Refs: 21
ISSN: 1434-5161 CODEN: JHGEFR
COUNTRY: Japan
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 022 Human Genetics
025 Hematology
LANGUAGE: English
SUMMARY LANGUAGE: English
AB The Rh blood group is the most polymorphic human blood group system, and is clinically significant in transfusion **medicine**. **Individuals** are classified as Rh-positive and Rh-negative depending on the presence or absence of the D antigen on the red cell surface. The RhD-negative trait could be generated by multiple genetic mechanisms, which have been shown to be ethnic group- dependent. In this study, we evaluated the status of seven RHD-specific exons (exons 3, 4, 5, 6, 7, 9, and 10) and RH intron 4 in 119 Chinese blood donors, using the **sequence**-specific primers polymerase chain reaction (SSP-PCR). Of the 87 individuals who were RhD-negative, 52 with the ce/ce, ce/cE, or Ce/ce genotype (60%) lacked the above seven RHD exons; 22 with the Ce/Ce or Ce/ce genotype (25%) had all the RHD exons examined; 13 with the Ce/ce genotype (15%) carried at least one RHD exon. Antigen association analysis suggested the existence of a novel class of RhD-negative associated haplotypes in the Chinese, tentatively denoted D(nf)Ce. The D(nf)Ce haplotype consisted of a normal RHCe allele and a nonfunctional RHD **gene**, which vary depending on the structure of the RHD **gene**. Among the RhD-negative Chinese, the estimated frequencies of the dce, dCe, and D(nf)Ce haplotypes were 0.7500, 0.0465, and 0.2035, respectively. No statistically significant deviation from Hardy- Weinberg equilibrium was observed using this genetic model.